BGI, Children's Hospital of Philadelphia launch the 1,000 Rare Diseases Project

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BGI, the world's largest genomics organization, and The Children's Hospital of Philadelphia (CHOP) today announced that they have jointly initiated the 1,000 Rare Diseases Project with the aim of accelerating the discovery of genetic variants underlying rare diseases.

The project will employ integrative genomic approaches and innovative analysis pipelines, laying a solid genetic foundation for future clinical diagnosis and treatment. The project primarily focuses on sequencing 1,000 rare diseases, including ones that affect both children and adults. Under the collaboration agreement, BGI and CHOP will use next-generation sequencing (NGS) technologies to analyze well characterized DNA samples from patients and families with single-gene inheritance patterns.

Rare diseases usually refer to either life-threatening or chronically debilitating diseases - most of which are inherited - that individually affect a small percentage of the population, but in total, have a large impact, affecting approximately 1 in every 12 newborn children. It is estimated that there are between 7,000 and 8,000 known rare diseases in the world. However, patients with rare diseases are currently a significantly underserved population, with insufficient social and medical support, because the small numbers of patients for any given disease makes it challenging to recoup the medical investments in research and development of therapies.

NGS technologies have revolutionized life sciences, allowing researchers to efficiently identify the genetic variants underlying rare diseases through whole-exome or whole-genome sequencing. "Experimental results show that the genome-wide sequencing approaches we are currently pursuing are more cost-effective and efficient than previous linkage-mapping and candidate gene methods. This is partly due to the fact that genome-wide sequencing approaches make it possible to pinpoint the cause of many rare diseases using much smaller numbers of samples," said Xun Xu, Deputy Director of BGI.

"The BGI/CHOP collaboration is an ideal partnership," said Hakon Hakonarson, M.D., Ph.D., Director of the Center for Applied Genomics at CHOP and co-Director of BGI@CHOP Joint Genome Center; "It brings together the unique strengths of two world-class institutions, combining BGI's robust capabilities and expertise in NGS and bioinformatics analysis with CHOP's extensive biobanking and clinical and translational expertise. This will undoubtedly facilitate rapid and accurate diagnosis of rare diseases and lead to new therapeutic interventions," said Hakonarson.

"We welcome this opportunity to work with one of the largest and most prestigious children's hospitals in the world," said Professor Jian Wang, President of BGI, "We would like to enhance rare diseases research through collaborative projects with researchers worldwide to help conquer rare diseases and improve the health and quality of life of those living with a rare condition."

Provided by BGI Shenzhen